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Review Article

A systematic review of ABCG8 mutation and sitosterolemia

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Abstract: Background: Sitosterolemia is a rare inherited condition caused by elevated levels of plant sterols in the plasma, characterized by mutations in ABCG5 and ABCG8 genes. A scarce occurrence in this condition are hematological abnormalities such as hemolytic anemia, stomatocytosis, and macrothrombocytopenia. We conducted a meta-analysis and systematic review to answer these questions regarding patients who have hemolytic anemia and ABCG8 mutation. Methods: 13 reports were shortlisted for the final analysis (Observational studies-6, case series-4, case reports-3). Descriptive statistics were utilized to study the patient characteristics. Results: From the 13 reports that we found in available literature, we identified 19 cases of ABCG8 mutation and anemia. From the random-effects proportions model, the chance of this event occurring among patients with sitosterolemia was 6.8% [0.068, 95% Confidence Interval (CI) 0.016-0.120, P=0.010] (I^2 24.68%) (14/145). Thrombocytopenia and stomatocytosis were frequently reported. Splenomegaly and xanthomas were other common associations. Conclusions: To the best of our knowledge, we provide the first report of the prevalence of anemia, specifically in patients with sitosterolemia caused by a mutation in the ABCG8 gene. At 6.8%, this is an extremely rare occurrence in an already infrequent disease.

Keywords: ABCG8, ABCG5, sitosterolemia, hemolytic anemia, xanthoma

Introduction

Sitosterolemia is a rare inherited condition caused by elevated levels of plant sterols in the plasma. Homozygous and compound heterozygous mutations in ABCG5 and ABCG8 genes characterize the genetic basis of the condition [1, 2]. The condition is very rare, such that information about this in literature is limited to case reports, case series' and translational- observational studies [3-5]. The ABCG5 and ABCG8 genes code for ABCG5 and ABCG8 protein respectively which serve as efflux pumps eliminating plant-based sterols into the gut lumen for excretion. Mutation in these genes leads to impairment in this process leading to absorption of plant sterols into blood. This elevation of plant sterols in blood is termed as sitosterolemia. A scarce occurrence in this already infrequent condition is hematological abnormalities such as hemolytic anemia, stomatocytosis, and macrothrombocytopenia.

Baseline characteristics of the disease such as frequency of occurrence, demographic details of patients, and degree of anemia are not readily available [6]. We conducted a meta-analysis and systematic review to answer these questions regarding patients who have hemolytic anemia and ABCG8 mutation.

Methodology

The systematic review was done as per the guidelines set forth by Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA guidelines) [7]. A systematic search was done with controlled vocabulary encompassing the terms ABCG8, Sitosterolemia, Phytosterolemia, Anemia, Hemolysis, hemolytic anemia, Autoimmune hemolytic anemia, Cold agglutinin disease, and Coombs was done. Databases including Pubmed, Embase, Scopus, and Cochrane were queried. No restrictions on date, language, status, or outcomes were applied. Initial search resulted in 627

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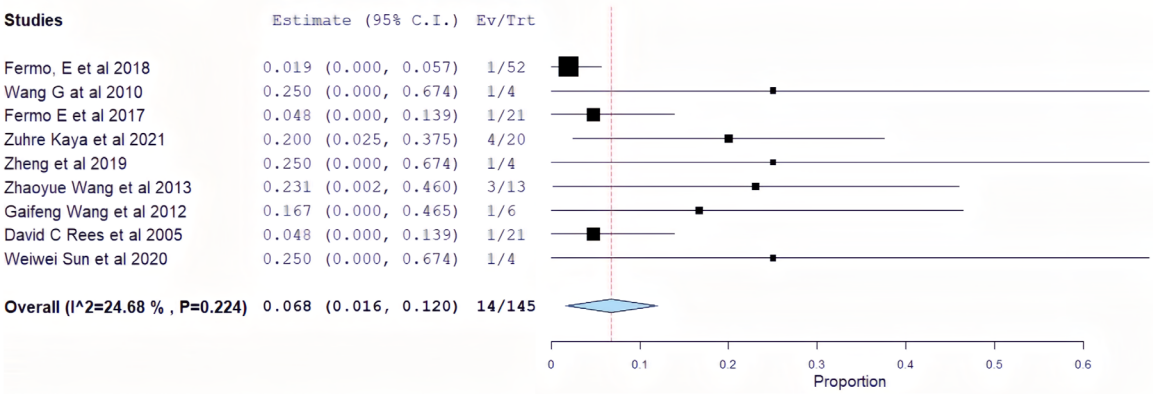

Figure 1. Forest plot depicting pooled proportions ratio of anemia and ABCG8 mutation occurring in patients with sitosterolemia.

(Pubmed - 33, Embase - 102, Scopus - 474, Cochrane - 18) entries. The search was done from inception to July 20, 2023. The records were imported into Mendeley, and duplicates were removed leaving us with 532 records. The records were imported to Rayyan https://www.rayyan.ai/.

The titles were screened independently by 2 reviewers (PAK and DD) and conflicts arising between the reviewers were resolved by a 3rd reviewer (KG). Inclusion criteria involved studies pertaining to patients with sitosterolemia, ABCG8 mutation and anemia of any cause or severity. Both adult and pediatric reports were included. Studies associated with ABCG5 mutation alone or lacked anemia were excluded. 13 reports were shortlisted for the final analysis (Observational studies-6, case series-4, case reports-3).

A data collection sheet was made and parameters like study features, data pertaining to the clinical course, patient characteristics, and adverse events were collected. Two authors (PAK, DD) independently collected and extracted the data. The data was reviewed by two other authors (DP and KG) for potential discrepancies, and if present, were resolved. Quality was verified using a scale based on the Newcastle-Ottawa Scale for meta-analysis [8]. The scale consisted of 7 questions, each of which was awarded 1 or 2 points based on the question, amounting to a maximum possible 9 points per study. We scored the questions in the scale based on the outcomes that our study aimed to find. We stratified the score as ≥7, 3-6, and <2 to depict good, fair, and poor-quality studies, respectively. 2 authors (DP and PAK) independently assessed the quality of the studies. The senior author reviewed the final quality assessment table and resolved discrepancies if there were any found. This has been included as a supplement.

Descriptive statistics were utilized to study the patient characteristics. The proportion of patients having both anemia and any ABCG8 mutation among all cases of sitosterolemia was determined using the DerSimonian-Laird random effects model with OpenMeta http://www.cebm.brown.edu/openmeta/.

The supplement contains the PRISMA diagram, Newcastle-Ottawa quality assessment table and the search strategy used.

Results

From the 13 reports we found in available literature, we identified 19 cases of ABCG8 mutation and anemia. From the random-effects proportions model, the chance of this event occurring among patients with sitosterolemia was 6.8% [0.068, 95% Confidence Interval (CI) 0.016-0.120, P=0.010] (I^2 24.68%) (14/145) (**Figure 1**). Various characteristics of the cohort analyzed, including the different ABCG8 mutations, are summarized in **Table 1**. The mean age of the patients was 26 years (Age range: 0.1-61 years) and included neonates, pediatric, and adult patients. There were 7 females and 7 males among those whose sex was reported. 9/13 studies documented elevated sterols, while 8/13 clearly stated hemolysis as the cause of anemia. The hemoglobin ranged

Table 1. Summary of various characteristics of the cohort analyzed, including the different ABCG8 mutations

S. No	Study	Year	Study Type	No. of Patients	Age (yrs)	Sex	ABCG8 Mutation	Sterol↑	Anemia Type	Hb (g/dL)	MCV (fL)	Platelet Abnormalities	Treatment	Anemia Response	Other Features
1	Garg et al	2014	Case report	1	58	F	Homozygous p.W536*	Yes	Normocytic	8.2	88.2	Thrombocytopenia (61 K)	-	-	Splenomegaly
2	Fermo et al	2018	Obs. study	1 (52)	-	-	Unspecified	-	Congenital hemolytic	-	-	-	-	-	-
3	Wang G et al	2010	Case series	1 (4)	-	-	del1938C-1939G/ ins1938T	Yes	Hemolytic	-	-	Macrothrombocytopenia, stomatocytosis	-	-	Splenomegaly, xanthomas
4	Fermo et al	2017	Obs. study	1 (21)	-	-	Unspecified	-	Congenital hemolytic	-	-	-	-	-	-
5	Kaya et al	2021	Obs. study	4 (20)	3-16	F-2, M-2	Homo/het p.A588P	Yes	Hemolytic	11-12	-	Macrothrombocytopenia	-	-	Splenomegaly
6	Wang W et al	2018	Case report	1	1.5	-	Compound: p.R164X, delTTC	Yes	Unspecified	10.1	-	Stomatocytosis	Ezetimibe	-	-
7	Zheng et al	2019	Case series	1 (4)	11	F	Compound: p.L228P, p.G575D	Yes	Normocytic	8.8	94.3	Thrombocytopenia (3 K)	Ezetimibe	Yes	ITP, epistaxis
8	Zhaoyue Wang et al	2013	Obs. study	3 (13)	23, 57, 61	M-3	Multiple exon mutations	Yes	Unspecified	10.2-12.6	-	Thrombocytopenia (14-39 K)	-	-	Splenomegaly, xanthomas
9	Gaifeng Wang et al	2012	Obs. study	1 (6)	43	F	del43683-43724, ins43866T	Yes	Hemolytic	11.1	-	Macrothrombocytopenia, stomatocytosis (78 K)	Splenectomy	Yes	Splenomegaly, xanthomas
10	Seabury et al	2023	Case series	2 (2)	0.1	F-1, M-1	p.G18R	-	Hemolytic	6.1, 11	-	-	Phototherapy, folic acid	Yes	-
11	Melenotte et al	2014	Case report	1	59	F	Homozygous p.Gln302*	-	Hemolytic	8	-	Thrombocytopenia (63 K)	Simvastatin	No	Splenomegaly, xanthomas, vasculopathy
12	Rees et al	2005	Obs. study	1 (21)	24	F	W361X (1083G>A)	Yes	Normocytic	7.7	90.5	-	-	-	-
13	Sun et al	2020	Case series	1 (4)	8	M	p.Ser473Ter, p.Gly512Arg	Yes	Hemolytic	11.8	101.6	Thrombocytopenia (60 K)	Antioxidants	-	Splenomegaly, cardiac enlargement

between 6.1 to 12.6 g/dl with a mean of 10.2 g/dl. Thrombocytopenia and stomatocytosis were frequently reported. Splenomegaly and xanthomas were other common associations. One example demonstrated an improvement in anemia after ezetimibe therapy, while splenectomy and supportive care was utilized in a couple of other reports to treat hemolysis. A summary of all studies included is presented in **Table 2**.

Discussion

Genetic defects in either ABCG5 and/or ABCG8 result in a rare inherited disease characterized by elevated levels of plant sterols in blood to 20 to 50 times the upper limit of normal, termed 'sitosterolemia'. These genes code for ABCG5 and ABCG8 proteins, heterodimer molecules known as sterolins, which are present on enterocytes and hepatocytes. These proteins function as efflux pumps, pumping out 90-99% of the plant sterols absorbed from diet in enterocytes or from hepatocytes into bile canaliculi; for excretion into the gut lumen. Genetic defects resulting in 75% or greater reduction in activity of these pumps results in pathological accumulation of plant sterols causing sitosterolemia [9, 10]. Sitosterolemia is a possible misnomer, named as such due to 'sitosterol' being the first plant sterol found to be elevated when this condition was initially described, however there is an increase of all plant sterols in blood i.e. a xenosterolemia [10].

Xenosterolemia presents characteristically with hypercholesterolemia, xanthomas, premature atherosclerosis, hemolytic anemia and macrothrombocytopenia with presence of stomatocytes. Occasionally, liver disease is also associated with this condition. In this condition, the hypercholesterolemia has been described as inversely proportional to age in its severity i.e. more severe in younger people and milder in older population [10].

'Sitosterolemia' characteristically does not respond to statins. The treatment of choice is NPC1L1 inhibitor ezetimibe [3, 4, 11]. It also responds very well to a low-fat diet. With advances in next generation sequencing technologies, multiple genetic variants of the ABCG5 and ABCG8 genes have been identified which indicates that the prevalence of sitosterolemia might be higher than was initially suggested with studies showing a general population prevalence of 5 in every 10,000 people in a US based study. Around 4% of patients with a diagnosis of 'familial hypercholesterolemia' have elevated levels of plant sterols [12]. It is thus important to maintain a high index of suspicion for the diagnosis of this condition. A regular enzyme-based lipid panel may not adequately differentiate between cholesterol and plant sterols. Another group for consideration are those without a response to adequate statin therapy. Specialized blood tests such as gas-chromatography mass spectrometry (GC-MS) or high-pressure liquid chromatography (HPLC) can be used to detect elevated levels of plant sterols. Genetic testing for ABCG5 and ABCG8 mutation is available to confirm the diagnosis. The initiation of ezetimibe and adherence to a low-fat diet has reportedly been associated with a good response including lowering of sterol levels in blood and reversal of xanthomas.

A complete blood count with differential can be very useful in patients with lab-reported dyslipidemia. The anemia associated with xenosterolemia is usually mild however the presence of anemia, evidence of hemolysis on peripheral smear and/or the presence of stomatocytes with macrothrombocytopenia can point towards a diagnosis of sitosterolemia. Concomitantly, splenomegaly, 'idiopathic' liver disease have also been associated with this condition and should raise clinical suspicion [4, 5, 14].

In 20-30% of adult patients with sitosterolemia, hemolytic anemia might be the solitary presenting feature of this condition [13]. Red blood cell (RBC) membranes comprise a lipid bilayer. This lipid bilayer is made up of cholesterol, sphingolipids and phospholipids amongst other constituents. Plant sterols or phytosterols exhibit a high degree of similarity with cholesterol. In sitosterolemia, there are abnormally elevated levels of plant sterols in the blood and these xenosterols are used instead of cholesterol in construction of the RBC membrane. These xenosterols fail to give the RBC membrane the structural integrity and durability given by cholesterol and this leads to stiffness and increased fragility of these cells ultimately leading to increased hemolysis.

Management of this hemolytic anemia is largely based around reducing the amount of xenos-

Table 2. Summary of reports included in this review

Study	Year	Study type	No of patients	Age	Sex	Elevated plant sterols	Treatment used	Other features
Garg et al [16]	2014	case report	1	58	F	Y	-	splenomegaly
Fermo et al [17]	2018	observational study	1	-	-	-	-	
Wang et al [18]	2010	case series	1	-	-	Y	-	splenomegaly, xanthomas
Fermo et al [19]	2017	observational study	1		-		-	
Zuhre Kaya et al [20]	2021	observational study	4	3, 12, 13, 16	M-2 F-2	Y	-	splenomegaly
Wang et al [21]	2018	case report	1	1.5	-	Y	Ezetimibe	
Zheng et al [5]	2019	case series	1	11	F	Y	Ezetimibe	ITP
Zhaoyue Wang et al [22]	2014	observational study	3	23, 57, 65	M-3	Y	-	splenomegaly, xanthomas
Gaifeng Wang et al [23]	2012	observational study	1	43	F	Y	splenectomy	splenomegaly, xanthomas
Seabury et al [24]	2023	case series	2	0.1	M-1 F-1	-	phototherapy, folic acid, supportive care	
Clea Melenotte et al [25]	2014	case report	1	59	F	-	simvastatin	splenomegaly, xanthomas
David Rees et al [26]	2005	observational study	1	24	F	Y	-	
Weiwei Sun et al [27]	2020	case series	1	8	M	Y	reduced glutathione and serum sarmentosum bunge granules	splenomegaly, cardiomegaly

terols present in the blood. As mentioned above, a low fat diet is helpful. Specifically avoiding foods with a high concentration of phytosterols is important such as seeds, nuts, oils derived from seeds and nuts and vegetable oils. Ezetimibe is an inhibitor of the NPC1L1 transporter thus acts to reduce the quantity of absorbed plant sterols from diet. It has reports of good efficacy in lowering levels of phytosterols in blood [4, 11] but so far has only shown to be effective for regulating function of newly developed red blood cells after reduction of blood xenosterol levels and does not appear to be effective in altering hemolysis of existing red blood cells. Bile-acid resin cholestyramine has been used as an adjunct to ezetimibe on occasion, in sitosterolemia management.

Currently, there is limited availability of clinical testing methods to detect plant sterols, gas chromatography being a relatively reliable method to accurately do so at this time [15]. As methods of identifying ABCG5/8 genetic variants or detecting phytosterols in blood become more easily accessible and widely available, it is expected that the rate of diagnosis of sitosterolemia will increase. This remains important as the first choice and most widely used management for dyslipidemia in the general population is statin-therapy, a class of medications that sitosterolemia is unlikely to respond to. With increased uncovering of this condition and research to further understanding of its pathophysiology, we might expect novel research into effective management strategies in the near future.

Conclusion

Sitosterolemia is a rare inherited genetic condition caused by mutation in ABCG5 and ABCG8 genes leading to elevated levels of plant sterols in blood. Features associated with sitosterolemia include hypercholesterolemia usually refractory to statins, xanthomas, hemolytic anemia and macrothrombocytopenia. This condition may not be evident on regular lipid panels making diagnosis challenging. Interventions like a low-fat diet and ezetimibe have demonstrated a good response making early diagnosis and adequate intervention crucial thus a high index of suspicion should be maintained in a patient presenting with elevated cholesterol and hemolytic anemia and/or macrothrombocytopenia.

As our understanding and identification of sitosterolemia improves, it is hoped that confirmatory diagnostic tools will be more accessible in the future.

Disclosure of conflict of interest

None.

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